

Organic Acid Disorder (OAD) Screening Fact Sheet for Health Care Providers  
Newborn Screening Program of the Oklahoma State Department of Health

**What are the characteristics of Organic Acid Disorders (OA)?**

- Autosomal recessive genetic conditions. Most infants are born to parents who are both unknowingly asymptomatic carriers and have NO known history of an Organic acid disorder in the family.
- The incidence of OA's ranges from 1/75,000 – 1/100,000.
- Symptoms vary by disorder. These disorders can lead to metabolic crisis, especially in infants and children. This crisis can lead to seizures, respiratory failure, cardiac arrest and death. Crisis survivors may experience significant developmental disabilities. Some infants may present in the neonatal period without any symptoms
- Symptoms of a metabolic crisis can be triggered by fever, infection or illness.
- Treatment involves a special diet managed by a metabolic specialist and a metabolic dietician, frequent feedings, and special care during times of illness or stress.

**What is the screening methodology for Organic Acid disorders?**

An acylcarnitine profile by Tandem Mass Spectrometry (MS/MS) is performed on each filter paper. Primary and secondary analytes are simultaneously tested on each filter paper. Primary analyte results are used to establish results requiring follow-up. All out-of-range primary analyte results require follow-up. Secondary analyte are used in conjunction with primary analyte results to assign risk. Elevations of the secondary analytes are reported as "not consistent with OA" if the primary analyte is in range.

**What are the follow-up needs?**

The follow-up program will provide detailed guidance on needed actions. The following metabolic specialists have approved all recommendations:

- Integris Pediatric Specialty Clinic, Inborn Error of Metabolism (IEM) Clinic
  - Geneticist pager: (405) 630-3794
- OU Children's Physicians – Genetics Clinic
  - Page Operator: (405) 271-3636
- Metabolic Nurse Specialist – (405) 271-8001 ext 42074

**What is my role in screening?**

If you are listed as the infant's planned health care provider on the filter paper requisition, you are required by the *Newborn Screening* to initiate follow up activities.

Summary of out of range OA Analytes and follow-up recommendations.				
Primary Marker (μmol/L)	Secondary Analyte (μmol/L)	Potential Disorder	Risk*	Recommendation
C3 ≥ 7.21	C3/C2 <0.25	PROP/MUT/CBL-C,D/CBL-A,B	LR	Repeat Filter paper. Consult metabolic specialist as needed.
C3 ≥ 7.21	C3/C2 ≥0.25	PROP/MUT/CBL-C,D/CBL-A,B	HR	Immediate referral to metabolic specialist. Confirmatory testing
C3DC 0.44-0.59	NONE	MAL	LR	Repeat Filter paper. Consult metabolic specialist as needed
C3DC ≥ 0.60	NONE	MAL	HR	Immediate referral to metabolic specialist. Confirmatory testing
C4 1.27 - 1.99	C4/C2 <0.06	IBG	LR	Repeat Filter paper. Consult metabolic specialist as needed.
C4 1.27 - 1.99	C4/C2 ≥0.06	IBG	HR	Immediate referral to metabolic specialist. Confirmatory testing
C4 ≥ 2.00		IBG	HR	Immediate referral to metabolic specialist. Confirmatory testing
C5 0.87-2.99	C5/C2 <0.05	IVA / 2MBG	LR	Repeat Filter paper. Consult metabolic specialist as needed.
C5 0.87-2.99	C5/C2 ≥0.05	IVA / 2MBG	HR	Immediate referral to metabolic specialist. Confirmatory testing
C5 ≥3.00		IVA / 2MBG	HR	Immediate referral to metabolic specialist. Confirmatory testing
C5:1 ≥0.10	C5OH+C4DC <0.80	βKT	LR	Repeat Filter paper. Consult metabolic specialist as needed.
C5:1 ≥0.10	C5OH+C4DC ≥0.80	βKT	HR	Immediate referral to metabolic specialist. Confirmatory testing
C5DC ≥0.46	C5DC/C8 <4.60 & C5DC/C16 <0.20	GA1	LR	Repeat Filter paper. Consult metabolic specialist as needed.
C5DC ≥0.46	C5DC/C8 ≥4.60	GA1	HR	Immediate referral to metabolic specialist. Confirmatory testing
C5DC ≥0.46	C5DC/C16 ≥0.20	GA1	HR	Immediate referral to metabolic specialist. Confirmatory testing
C5OH+C4DC ≥0.80	C6DC <0.27	HMG/3MCC/3MGA/MCD/2M3HBA	LR	Repeat Filter paper. Consult metabolic specialist as needed.
C5OH+C4DC ≥0.80	C6DC ≥0.27	HMG/3MCC/3MGA/MCD/2M3HBA	HR	Immediate referral to metabolic specialist. Confirmatory testing
C5OH+C4DC ≥2.0	C6DC <0.27	HMG/3MCC/3MGA/MCD/2M3HBA	HR	Immediate referral to metabolic specialist. Confirmatory testing

Cut off values may change. Please contact the OK NBS Program for clarification if needed. More information available on OSDH website or call (405) 271-6617.

\*HR-High Risk  
\*LR-Low Risk